

Whole exome sequencing identified novel *CRB1* mutations in Chinese and Indian population with autosomal recessive retinitis pigmentosa

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Supplementary information file

Supplementary information file includes one table (table S1) and two figures (figure S1 and figure S2).

Table S1

The next-generation sequencing results of the familial samples and the 100 unrelated cases.

	total reads that pass QC	Initial mappable reads	On-target reads	% Coverage of target regions(more than 10×)	Mean read depth of target regions
IV:2 in RP-2236	46,130,930	45,962,518	34,249,841	94.6%	57.0
II:1 in RP-IC-90	53,474,296	53,303,980	40,791,416	96.3%	68.6
100 sporadic samples(average)	49,479,221 ± 9819444.07	49,295,539 ± 9779248.38	37,721,369 ± 4228047.62	95.2% ± 1.32%	62.3 ± 5.32

Figure Legends

Figure S1. The full field ERG of patient IV:2. The full field ERG test of patient IV:2 showed almost no response under the scotopic condition and weak response under the photopic condition. The Incubation period of b wave prolonged and its amplitude decreased. This test indicated significant loss of function of both rods and cones.

Figure S2. Fundus photographs of other patients. (A) and (B) Fundus photography of patient IV:3 showed pale optic papilla, thinning of retinal vessels and pigmentation all over the retina in each eye. (C) and (D) Fundus photography of patient IV:4 showed unclear boundary of optic papilla, thinning of retinal vessels as well as pigment disorder and deposition in both eyes.

Fig S1

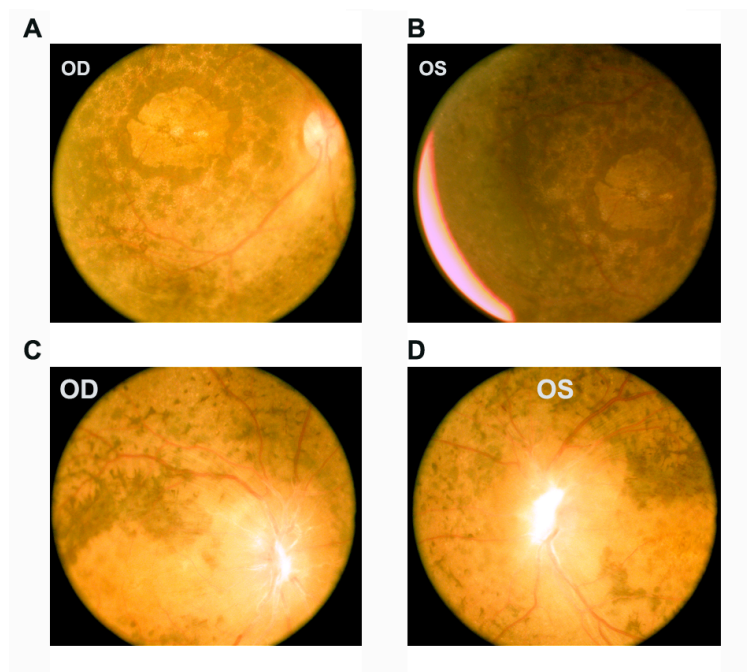


Fig S2

